SFRP4 gene

secreted frizzled related protein 4

Normal Function

The *SFRP4* gene provides instructions for making a protein called secreted frizzled-related protein 4 (SFRP4). This protein blocks (inhibits) a process called Wnt signaling. Wnt signaling plays an important role in the development of several tissues and organs throughout the body. In particular, regulation of this signaling process by SFRP4 is critical for normal bone development and remodeling. Bone remodeling is a normal process in which old bone is broken down and new bone is created to replace it. The SFRP4 protein also plays a role in the development of fatty (adipose) tissue.

Health Conditions Related to Genetic Changes

Dupuytren contracture

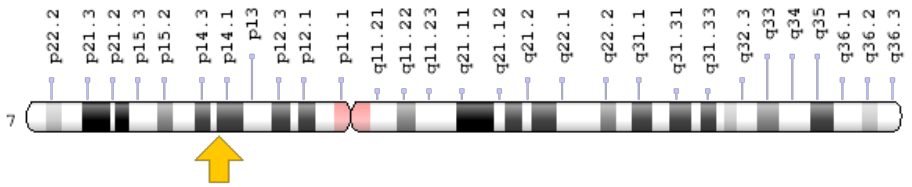
Pyle disease

At least four *SFRP4* gene mutations have been found in individuals with a bone disorder called Pyle disease. This condition is characterized by a bone abnormality in which the ends (metaphyses) of the long bones in the arms and legs are abnormally wide, resembling a boat oar or paddle. Other bones may also be abnormal in Pyle disease, including the collar bones (clavicles), ribs, and bones in the fingers and hands. The *SFRP4* gene mutations are thought to lead to production of an abnormally short SFRP4 protein with impaired function, or they result in no SFRP4 protein production at all. Studies suggest that loss of functional SFRP4 dysregulates Wnt signaling, which disrupts normal bone development and remodeling. Abnormal bone formation leads to the characteristics of Pyle disease.

Chromosomal Location

Cytogenetic Location: 7p14.1, which is the short (p) arm of chromosome 7 at position 14.1

Molecular Location: base pairs 37,905,932 to 37,916,923 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- frizzled protein, human endometrium
- FRP-4
- FRPHE
- PYL
- secreted frizzled-related protein 4 precursor
- secreted frizzled-related protein 4; secreted frizzled-related protein 4
- sFRP-4

Additional Information & Resources

Educational Resources

- Endotext (2000): Bone Remodeling and its Relationship to Bone Quantity https://www.ncbi.nlm.nih.gov/books/NBK279134/#_epidem-patho-osteopo_toc-bone-remodeling-and-its-relationship-to-bone-quantity_
- Madame Curie Bioscience Database (2000): Secreted Frizzled-Related Protein (sFRPs) https://www.ncbi.nlm.nih.gov/books/NBK6536/#_A16295_

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SFRP4%5BTIAB%5D%29+OR+%28secreted+frizzled+related+protein+4%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- SECRETED FRIZZLED-RELATED PROTEIN 4
  http://omim.org/entry/606570

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/SFRP4ID42277ch7p14.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SFRP4%5Bgene%5D
- HGNC Gene Family: Secreted frizzled-related proteins
  http://www.genenames.org/cgi-bin/genefamilies/set/733
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10778
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6424
- UniProt
  http://www.uniprot.org/uniprot/Q6FHJ7

Sources for This Summary

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